

Familial cardiac screening

Camp Odayin Family Camp

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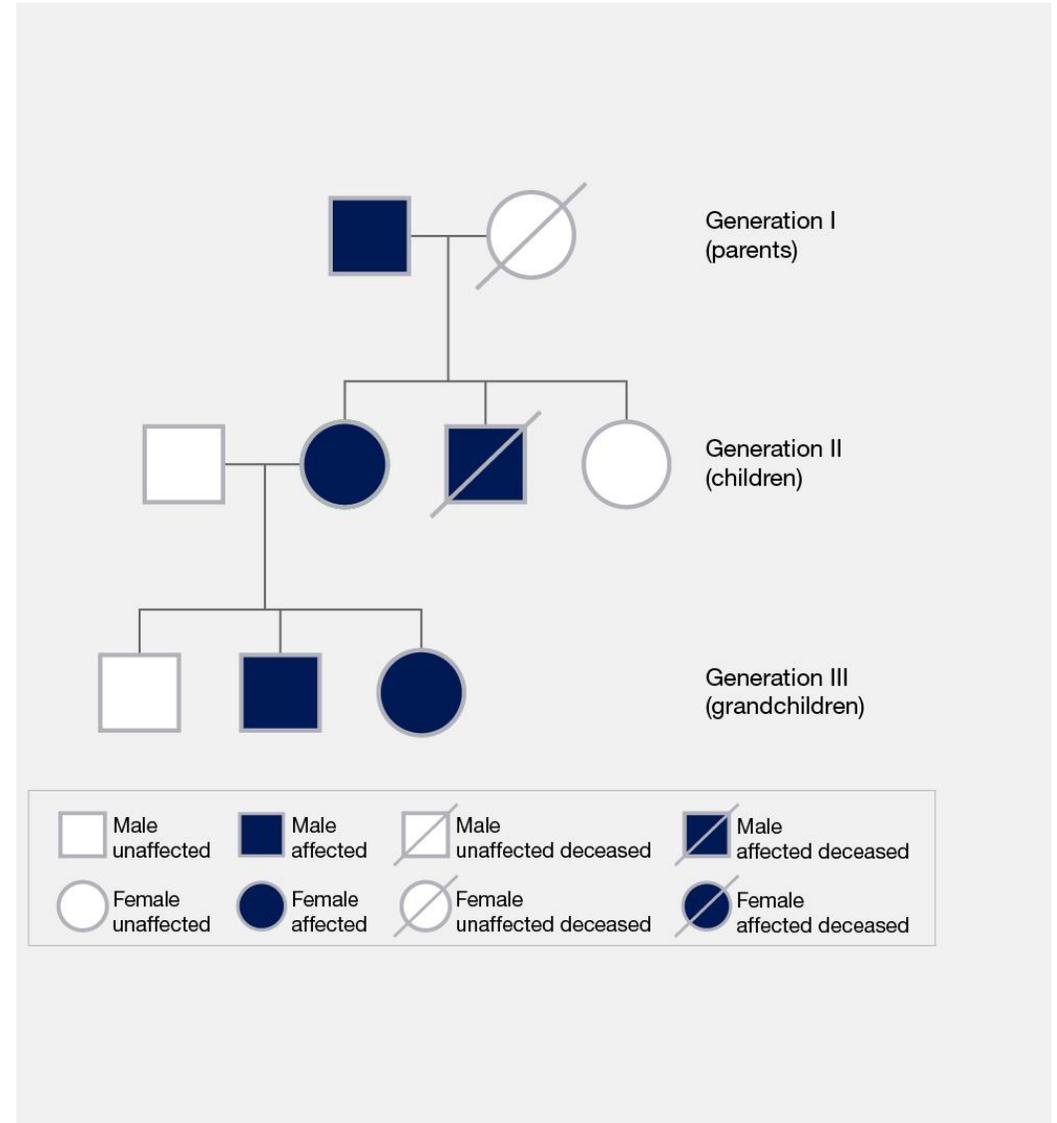
Disclosure: I'm NOT an expert

Familial Heart Screening

- Who?
 - What heart diseases qualify?
- Why?
 - What are we looking for? What can we find?
- What/How?
 - Echo? EKG?
- When?
 - Fetal echocardiogram?
 - Age?
 - One and done? Ongoing screening forever?

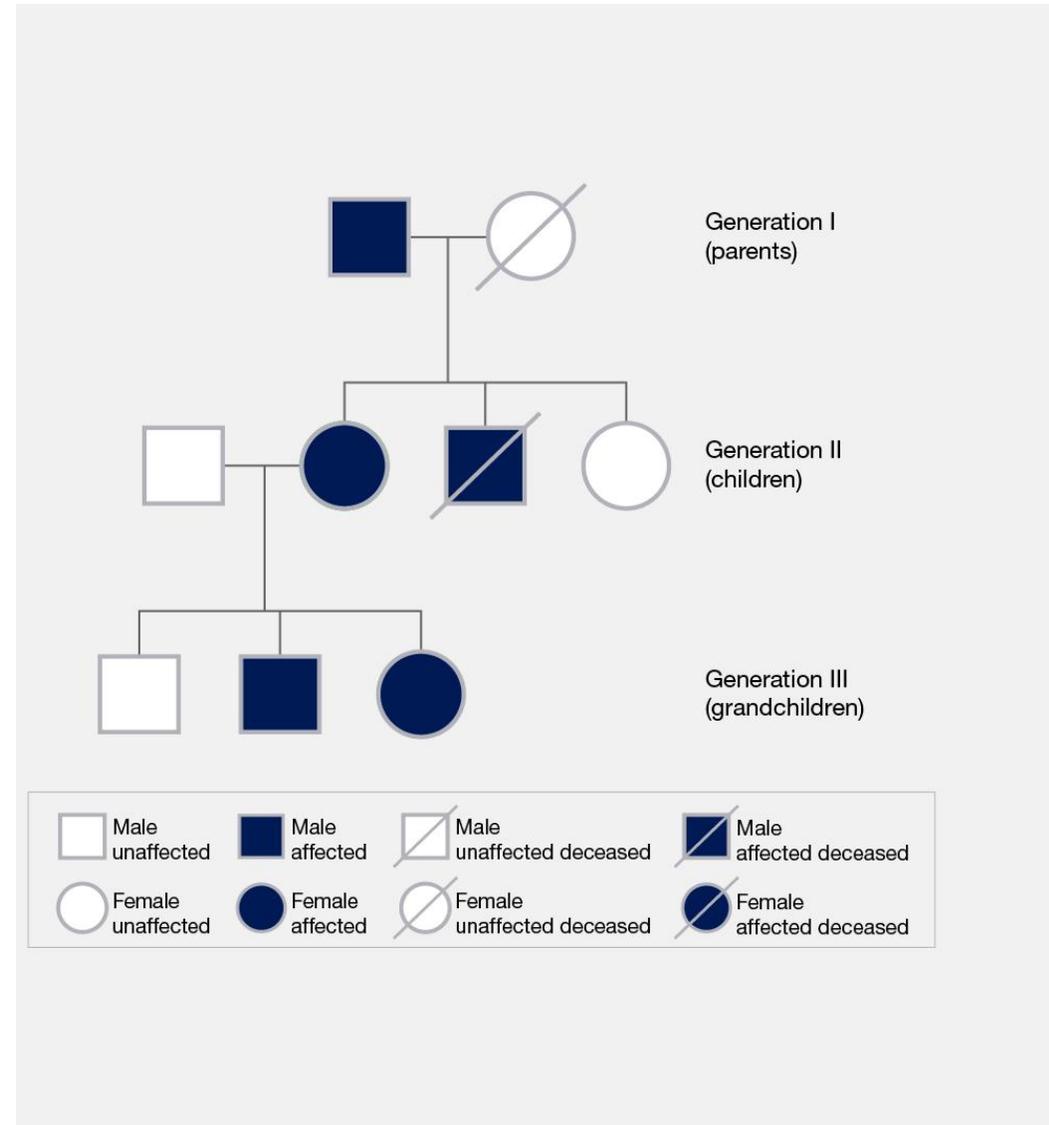
Who to screen

- Certain congenital heart diseases
 - Bicuspid aortic valve: 25% familial rate
- Cardiomyopathy
 - Up to 50% familial rate for autosomal dominant
 - Cardiac or genetic screening?
- Sudden cardiac death
 - Arrhythmia risks



Who to screen

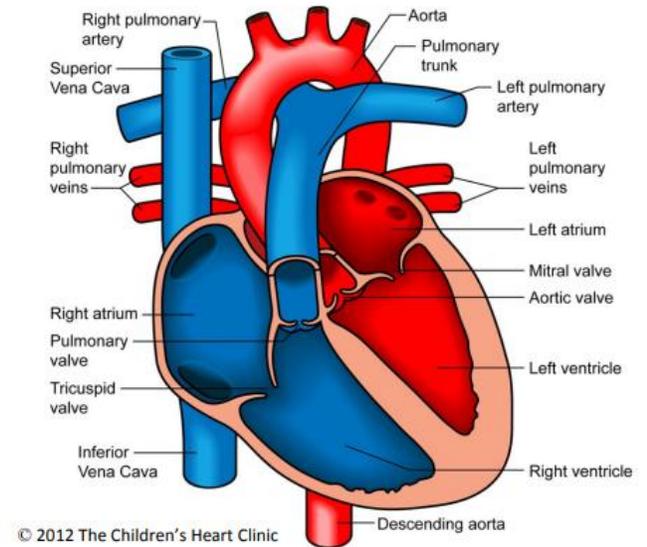
- First Degree family members of the affected person
- Siblings
- Parents
- Children (Yes, your daughters will get pregnant one day!)
- If new cases are found, the screening can expand
- e.g. if you screen positive, tell your siblings



What tests to screen with?

- Echocardiogram
 - Bicuspid aortic valve
 - Coarctation of the aorta
 - Hypoplastic left heart syndrome
 - Cardiomyopathy
- Fetal echocardiogram
 - Congenital heart disease
 - Cardiomyopathy
 - Certain arrhythmias (congenital heart block)
- EKG
 - Cardiomyopathy
 - Arrhythmias (long QT syndrome, etc)

Bicuspid Aortic Valve

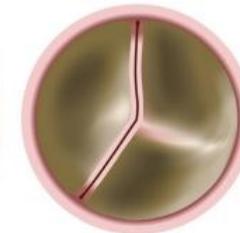


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Normal Aortic Valve



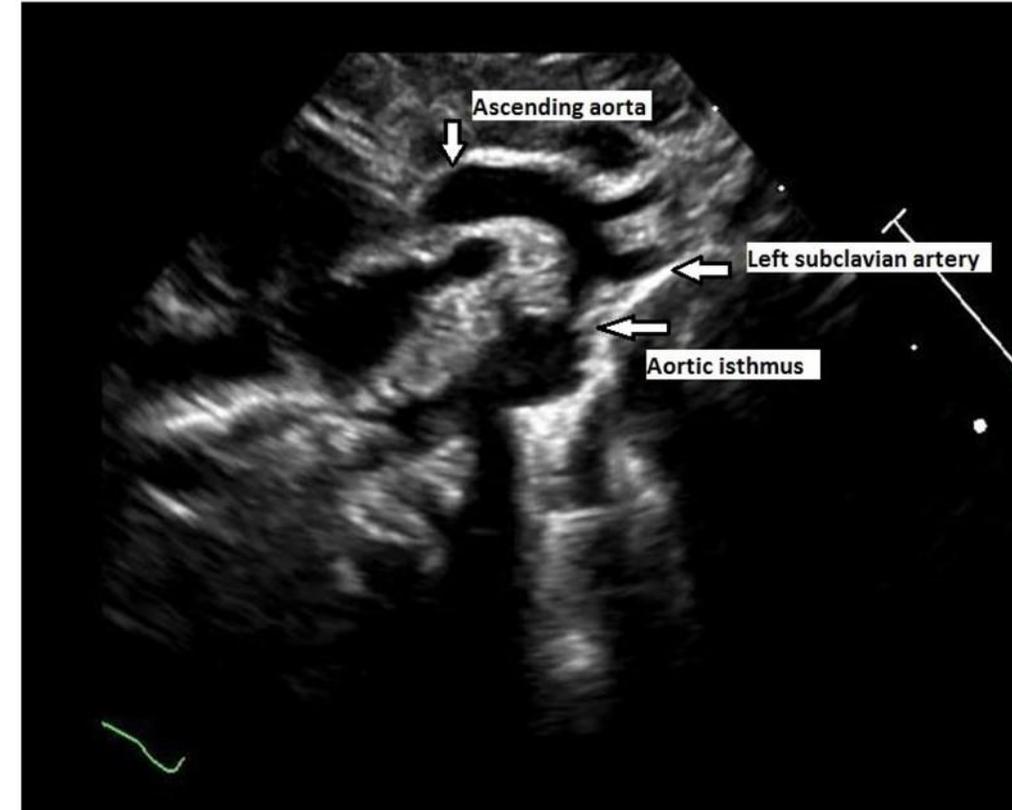
Bicuspid Aortic Valve



NOTES:

When to perform screening

- Before birth!
 - Early fetal echocardiogram at 16 weeks gestation
 - Gold standard at 20 weeks (same as anatomy scan/level II ultrasound)
- After birth
 - Echo usually recommended around 3-6 months old for best echo quality
- Ongoing screening
 - Cardiomyopathy or sudden cardiac death
 - EKG and echo every 3-5 years during childhood



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